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Severe Facial Clefting, Limbic Dermoid, Hypoplasia of the Corpus Callosum, and Multiple Skin Appendages: Severe Frontofacionasal "Dysplasia" or Newly Recognised Syndrome?

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We report on a child with severe midline facial cleft, bilateral cleft lip and palate, telecanthus, S-shaped palpebral fissures, limbic dermoid, midface hypoplasia, hypoplastic corpus callosum, and multiple skin appendages. This case may be an example of severe frontofacionasal "dysplasia" or a newly recognised syndrome.

KEY WORDS: facial cleft, cleft lip and palate

INTRODUCTION

The main clinical manifestations of frontofacionasal "dysplasia" (FFND) include severe midline facial defect, telecanthus, colobomatous eyelids, S-shaped palpebral fissures, absent ali nasi, and cleft lip and palate [Gollop, 1981, 1984]. We report on a child with severe midline facial defect, limbic dermoid skin appendages, and hypoplastic corpus callosum. The constellation of abnormalities in this child may represent severe FFND or a newly recognised syndrome.

CLINICAL REPORT

The patient is the 8th child of unrelated parents of Omani origin. All other sibs are normal. The father has mild hypertelorism. Birth weight was 3.5 kg.

At birth he was noted to have the following anomalies: broad forehead with laterally displaced eyebrows and telecanthus with S-shaped palpebral fissure with slight upward slant. The anterior hairline extended onto the eyebrows at the lateral margins. There was bilateral complete cleft lip and palate involving the hard and soft palate, defective left nostril, and macrostomia (Fig. 1). Eye examination showed a right limbic dermoid, but no other abnormalities. There were several skin appendages on the nose, right and left cheek, left preauricular region, and right ear lobe (Fig. 1). CT scan of the brain showed hypoplasia of the corpus callosum. CT scan of the facial bones showed huge bone defect in the maxilla and anterior part of the hard palate. The cleft extended into the soft palate and the palatopharyngeal muscles were not well developed (Fig. 2). In the nasal cavity, the septal cartilage was visible and a remnant of the maxillary bone was seen at the top of the nose. The choanae and the anterior and middle portion of the ethmoid bone was undeveloped. Biopsy of two of the skin appendages showed normal skin structure. Renal ultrasound was normal. Chromosomes were normal (46,XY).

DISCUSSION

Several manifestations in our patient are found in FFND (S-shaped palpebral fissures, limbic dermoid, facial cleft, and cleft lip-palate). FFND has autosomal recessive inheritance with extreme variability of the severity of abnormalities [Gollop, 1981, 1984]. Reardon et al. [1994] recently reported a case with FFND and severe eye involvement. They suggested that of the nine cases reported with this condition, diagnosis was definite in only four cases and the other five with mild involvement could represent a clinical variation of the phenotype or separate conditions.

The extent of clefting in this patient is much more severe than in the previously reported cases of FFND. Eyelid colobomata typical of FFND, were absent. In addition, the patient has partial agenesis of the corpus callosum and skin appendages which have not been reported in FFND. Furthermore, lack of consanguinity and the presence of seven sibs make autosomal recessive inheritance unlikely. The combination of abnormalities in this child may represent a previously undescribed syndrome.

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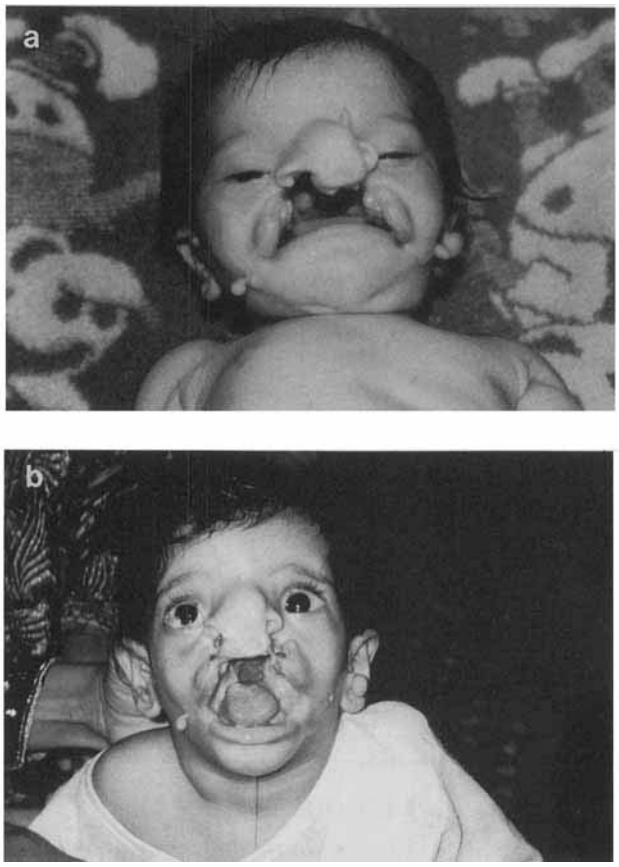

Fig. 1. Facial appearance at birth and age 1 year. Note S-shaped palpebral fissures, limbic dermoid of right eye, extensive bilateral cleft lip and palate, and multiple skin appendages.

Fig. 2. CT scan of facial bones. Maxilla has large central defect. Note palatal clefting, extreme protrusion of premaxillary segment, and hypoplasia of the palatal muscles.

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